	Type	L #	Hits	Search Text	DBs	Time Stamp	Comments	Error Definition	Errors
1	BRS	L1	85	arsacs	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:30			0
2	BRS	L2	9	spastin	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:34			0
3	BRS	L3	0	1 same mutat$3	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:37			0
4	BRS	L4	0	hybridiz$5 same 2	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:38			0
5	BRS	L5	0	sacsin	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:39			0
6	BRS	L6	2	1 same gene	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:40			0
7	BRS	L7	0	recombinant same 2	USPAT; US-PGPUB; EPO; DERWENT	2003/09/02 17:40			0

```
FILE 'MEDLINE' ENTERED AT 17:44:48 ON 02 SEP 2003

FILE 'CAPLUS' ENTERED AT 17:44:48 ON 02 SEP 2003
USE IS SUBJECT TO THE TERMS OF YOUR STN CUSTOMER AGREEMENT.
PLEASE SEE "HELP USAGETERMS" FOR DETAILS.
COPYRIGHT (C) 2003 AMERICAN CHEMICAL SOCIETY (ACS)

FILE 'BIOSIS' ENTERED AT 17:44:48 ON 02 SEP 2003
COPYRIGHT (C) 2003 BIOLOGICAL ABSTRACTS INC.(R)

FILE 'EMBASE' ENTERED AT 17:44:48 ON 02 SEP 2003
COPYRIGHT (C) 2003 Elsevier Science B.V.  All rights reserved.

FILE 'SCISEARCH' ENTERED AT 17:44:48 ON 02 SEP 2003
COPYRIGHT 2003 THOMSON ISI

FILE 'AGRICOLA' ENTERED AT 17:44:48 ON 02 SEP 2003

=> s spastin
L1          232 SPASTIN

=> s sacsin
L2           30 SACSIN

=> s l1 or l2
L3          257 L1 OR L2

=> s arsacs
L4           60 ARSACS

=> s l4 (p) mutat?
L5           25 L4 (P) MUTAT?

=> s l5 (p) l3
L6           12 L5 (P) L3

=> duplicate remove l6
DUPLICATE PREFERENCE IS 'MEDLINE, CAPLUS, BIOSIS, EMBASE, SCISEARCH'
KEEP DUPLICATES FROM MORE THAN ONE FILE?  Y/(N):n
PROCESSING COMPLETED FOR L6
L7             5 DUPLICATE REMOVE L6 (7 DUPLICATES REMOVED)

=> d l7 1-5 ibib abs

L7   ANSWER 1 OF 5  CAPLUS  COPYRIGHT 2003 ACS on STN
ACCESSION NUMBER:         2002:965729  CAPLUS
DOCUMENT NUMBER:          138:235578
TITLE:                    Autosomal recessive spastic ataxia of
                          Charlevoix-Saguenay (ARSACS/SACS)-no longer a local
                          disease
AUTHOR(S):                Richter, Andrea
CORPORATE SOURCE:         Service de Genetique Medicale, Hopital Sainte-Justine,
                          Departement de Pediatrie, Universite de Montreal,
                          Montreal, QC, Can.
SOURCE:                   Genetics of Movement Disorders (2003), 189-193.
                          Editor(s): Pulst, Stefan-M.  Elsevier Science: San
                          Diego, Calif.
                          CODEN: 69DIVT; ISBN: 0-12-566652-7
DOCUMENT TYPE:            Conference; General Review
LANGUAGE:                 English
AB   A review.  Autosomal recessive spastic ataxia of Charlevoix-Saguenay (
       ***ARSACS***  /SACS, OMIM 270550) originally described in 1978 is a clin.
     homogeneous form of early-onset familial disease with prominent myelinated
     retinal nerve fibers (Bouchard et al., 1991).  Over 300 patients were
     identified and most of their families originated in the
     Charlevoix-Saguenay region of Northeastern Quebec in Canada.  The
     frequency of several recessive diseases is increased in this region due to
     a founder effect caused by the settlement patterns of the late 17th to
     mid-19th centuries (Jette et al., 1991, Gauvreau et al., 1991,
     DeBraekeleer, 1991).  The gene carrier prevalence was estd. to be 1/22.
     (DeBraekeleer et al., 1993).  Patients present in early childhood with
     spastic gait ataxia.  The disease progresses rapidly in young adults and
     patients are wheelchair-bound by their fifth decade.  The  ***ARSACS***
     locus was mapped to chromosome region 13q11 by noting increased
     homozygosity for locus D13S787 in a genome-wide scan (Bouchard et al.,
     1998).  Following extensive genetic, phys., and transcript mapping
```

combined with directed sequencing, 2 ***mutations*** were detected in the ***Sacsin*** (SACS) g[redacted]e in ***ARSACS*** families [redacted]Richter et al., 1999, Engert et al., 1999, 2000). Both the single nucleotide deletion (g.6594delT, .DELTA.T) and the g.5254C > T (C > T) nonsense ***mutation*** cause the premature termination of the predicted ***sacsin*** protein. We calcd. disease allele frequencies using data from more than 125 Quebec ***ARSACS*** patients. Close to 94% of the disease alleles carried the .DELTA.T ***mutation*** , over 3% the C > T ***mutation*** . Interestingly close to 3% of the disease alleles carry unknown ***mutation*** (s), always in heterozygous form with .DELTA.T (Mercier et al., 2001). The sequencing of SACS is underway to identify these ***mutations*** . There are descriptions of recessive spastic ataxias clin. very similar to ***ARSACS*** in France (Chaigne et al., 1993), Tunisia (Mrissa et al., 2000), Spain (Pascual-Castroviejo et al., 2000), and Turkey (Gucuyener et al., 2001). The availability of family material in two of the studies permitted linkage anal. Results show that the disease linked to the SACS region on chromosome 13q in a large consanguineous kindred from Tunisia (Mrissa et al., 2000) and in two consanguineous families from Turkey (Gucuyener et al., 2001). These publications likely represent only the first few cases of recessive spastic ataxia where a diagnosis of ***ARSACS*** should be considered.
REFERENCE COUNT: 23 THERE ARE 23 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L7 ANSWER 2 OF 5 CAPLUS COPYRIGHT 2003 ACS on STN
ACCESSION NUMBER: 2001:300914 CAPLUS
DOCUMENT NUMBER: 134:324718
TITLE: Identification of the ***spastin*** gene associated with autosomal recessive spastic ataxia of Charlevoix-Saguenay (***ARSACS***) and diagnostic detection of ***mutations***
INVENTOR(S): Hudson, Thomas J.; Engert, James; Richter, Andrea
PATENT ASSIGNEE(S): Mcgill University, Can.; Hopital Sainte-Justine
SOURCE: PCT Int. Appl., 76 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2001029266	A2	20010426	WO 2000-US29130	20001020
WO 2001029266	A3	20020711		

W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CR, CU, CZ, DE, DK, DM, DZ, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG

PRIORITY APPLN. INFO.: US 1999-160588P P 19991020
AB Isolated spastin genes and fragments thereof, as well as Spastin proteins and fragments thereof are disclosed. Also disclosed are altered forms of spastin, as well as methods for the diagnosis and treatment of neurodegenerative disease.

L7 ANSWER 3 OF 5 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2003:381365 BIOSIS
DOCUMENT NUMBER: PREV200300381365
TITLE: ***ARSACS*** ; ***mutation*** detection and studies towards understanding the function of ***sacsin*** .
AUTHOR(S): Richter, A. M. (1); Mercier, J. (1); Engert, J. C.; LeBlanc, C. (1); Hudson, T. J.
CORPORATE SOURCE: (1) Centre de Recherche, Hopital Sainte-Justine, Universite de Montreal, Montreal, PQ, Canada: andrea.richter@umontreal.ca Canada
SOURCE: European Journal of Human Genetics, (2001) Vol. 9, No. Supplement 1, pp. P1443. print.
Meeting Info.: 10th International Congress of Human Genetics Vienna, Austria May 15-19, 2001 International Federation of Human Genetics Societies
. ISSN: 1018-4813.
DOCUMENT TYPE: Conference
LANGUAGE: English

L7 ANSWER 4 OF 5 MEDLINE on STN DUPLICATE 1
ACCESSION NUMBER: 2002062754 MEDLINE
DOCUMENT NUMBER: 21648525 PubMed ID: 11788093
TITLE: Rapid detection of the sacsin mutations causing autosomal recessive spastic ataxia of Charlevoix-Saguenay.
AUTHOR: Mercier J; Prevost C; Engert J C; Bouchard J P; Mathieu J; Richter A
CORPORATE SOURCE: Service de Genetique Medicale, Hopital Sainte-Justine, Departement de Pediatrie, Universite de Montreal, 3175 Cote Sainte Catherine, Montreal, Quebec, Canada H3T 1C5.
SOURCE: GENETIC TESTING, (2001 Fall) 5 (3) 255-9.
Journal code: 9802546. ISSN: 1090-6576.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200203
ENTRY DATE: Entered STN: 20020125
Last Updated on STN: 20020403
Entered Medline: 20020327
AB Autosomal recessive spastic ataxia of Charlevoix-Saguenay (***ARSACS*** ; MIM SACS 270550) is frequent in northeastern Quebec. Two causal ***mutations*** have been identified in the 11.7-kb single exon ***sacsin*** gene by sequence-based analyses. ***Mutation*** g.6594delT (DeltaT) was reported in 96% of the patients whereas a g.5254C --> T nonsense ***mutation*** has been observed only in 2 families. Here we report a reliable and inexpensive method to detect more than 95% of the ***ARSACS*** disease alleles from northeastern Quebec using allele-specific oligonucleotide (ASO) hybridization. This procedure is being incorporated into the diagnosis of ***ARSACS*** , as well as being used for carrier detection in at-risk families from northeastern Quebec.

L7 ANSWER 5 OF 5 CAPLUS COPYRIGHT 2003 ACS on STN DUPLICATE 2
ACCESSION NUMBER: 2000:114331 CAPLUS
DOCUMENT NUMBER: 132:263561
TITLE: ARSACS, a spastic ataxia common in northeastern Quebec, is caused by mutations in a new gene encoding an 11.5-kb ORF
AUTHOR(S): Engert, James C.; Berube, Pierre; Mercier, Jocelyne; Dore, Carole; Lepage, Pierre; Ge, Bing; Bouchard, Jean-Pierre; Mathieu, Jean; Melancon, Serge B.; Schalling, Martin; Lander, Eric S.; Morgan, Kenneth; Hudson, Thomas J.; Richter, Andrea
CORPORATE SOURCE: Montreal Genome Centre, McGill University Health Centre Research Institute, Montreal, QC, Can.
SOURCE: Nature Genetics (2000), 24(2), 120-125
CODEN: NGENEC; ISSN: 1061-4036
PUBLISHER: Nature America
DOCUMENT TYPE: Journal
LANGUAGE: English
AB Autosomal recessive spastic ataxia of Charlevoix-Saguenay (***ARSACS*** or SACS) is an early onset neurodegenerative disease with high prevalence (carrier frequency 1/22) in the Charlevoix-Saguenay-Lac-Saint-Jean (CSLSJ) region of Quebec. The authors previously mapped the gene responsible for ***ARSACS*** to chromosome 13q11 and identified two ancestral haplotypes. Here the authors report the cloning of this gene, SACS, which encodes the protein ***sacsin*** . The ORF of SACS is 11,487 bp and is encoded by a single gigantic exon spanning 12,794 bp. This exon is the largest to be identified in any vertebrate organism. The ORF is conserved in human and mouse. The putative protein contains three large segments with sequence similarity to each other and to the predicted protein of an Arabidopsis thaliana ORF. The presence of heat-shock domains suggests a function for ***sacsin*** in chaperone-mediated protein folding. SACS is expressed in a variety of tissues, including the central nervous system. The authors identified two SACS ***mutations*** in ***ARSACS*** families that lead to protein truncation, consistent with haplotype anal.
REFERENCE COUNT: 42 THERE ARE 42 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

=> d his

(FILE 'HOME' ENTERED AT 17:44:28 ON 02 SEP 2003)

```
     FILE 'MEDLINE, CAPLUS, BIOSIS, EMBASE, SCISEARCH, AGRICOLA' ENTERED AT
     17:44:48 ON 02 SEP 2003
L1          232 S SPASTIN
L2           30 S SACSIN
L3          257 S L1 OR L2
L4           60 S ARSACS
L5           25 S L4 (P) MUTAT?
L6           12 S L5 (P) L3
L7            5 DUPLICATE REMOVE L6 (7 DUPLICATES REMOVED)

=> duplicate remove l5
DUPLICATE PREFERENCE IS 'MEDLINE, CAPLUS, BIOSIS, EMBASE, SCISEARCH'
KEEP DUPLICATES FROM MORE THAN ONE FILE?  Y/(N):n
PROCESSING COMPLETED FOR L5
L8            8 DUPLICATE REMOVE L5 (17 DUPLICATES REMOVED)

=> s l8 not l7
L9            4 L8 NOT L7

=> d l9 1-4 ibib abs

L9   ANSWER 1 OF 4     MEDLINE on STN
ACCESSION NUMBER:   2003341319     MEDLINE
DOCUMENT NUMBER:    22755616   PubMed ID: 12873855
TITLE:              Phenotypic features and genetic findings in sacsin-related
                    autosomal recessive ataxia in Tunisia.
AUTHOR:             El Euch-Fayache Ghada; Lalani Irfan; Amouri Rim; Turki
                    Ilhem; Ouahchi Karim; Hung Wu-Yen; Belal Samir; Siddique
                    Teepu; Hentati Faycal
CORPORATE SOURCE:   Department of Neurology, National Institute of Neurology,
                    Tunis, Tunisia.
SOURCE:             ARCHIVES OF NEUROLOGY, (2003 Jul) 60 (7) 982-8.
                    Journal code: 0372436. ISSN: 0003-9942.
PUB. COUNTRY:       United States
DOCUMENT TYPE:      Journal; Article; (JOURNAL ARTICLE)
LANGUAGE:           English
FILE SEGMENT:       Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH:        200308
ENTRY DATE:         Entered STN: 20030723
                    Last Updated on STN: 20030807
                    Entered Medline: 20030806
AB   BACKGROUND: Autosomal recessive spastic ataxia of Charlevoix-Saguenay (
       ***ARSACS***  ) is a clinically homogenous disorder reported in Quebec
     caused by   ***mutations***   in the SACS gene (chromosome 13q12).
     Recently, we identified a Tunisian kindred demonstrating linkage to the
       ***ARSACS***   locus.  OBJECTIVE: To report clinical, neurophysiological,
     and nerve biopsy findings in patients with autosomal recessive cerebellar
     ataxia related to the SACS gene in Tunisia.  PATIENTS AND METHODS: Genetic
     linkage analysis of patients with early-onset autosomal recessive
     cerebellar ataxia allowed the identification of 4 families from which 18
     patients demonstrated linkage to the   ***ARSACS***   locus.  The patients
     were evaluated according to the International Cooperative Ataxia Rating
     Scale.  Peripheral nerve conduction, sensory evoked potentials, and nerve
     biopsy were performed in most patients.  RESULTS: The mean age at onset
     was 4.5 years.  The clinical phenotype was stereotyped and associated with
     a progressive cerebellar syndrome, a pyramidal syndrome with brisk knee
     reflexes, and Babinski sign and absent ankle reflexes.  The course of the
     disease varied among patients.  Sensory evoked potentials showed severe
     posterior column involvement.  Peripheral nerve investigations
     demonstrated axonal and demyelinating neuropathy.  Four   ***mutations***
     , 2 missense and 2 nonsense, were found.  CONCLUSION: In Tunisia,
     autosomal recessive cerebellar ataxia related to the SACS gene
     demonstrated a homogenous phenotype and heterogeneous allelic
       ***mutations***  .

L9   ANSWER 2 OF 4     MEDLINE on STN
ACCESSION NUMBER:   2000120709     MEDLINE
DOCUMENT NUMBER:    20120709   PubMed ID: 10655055
TITLE:                ***ARSACS***  , a spastic ataxia common in northeastern
                    Quebec, is caused by   ***mutations***   in a new gene
                    encoding an 11.5-kb ORF.
AUTHOR:             Engert J C; Berube P; Mercier J; Dore C; Lepage P; Ge B;
                    Bouchard J P; Mathieu J; Melancon S B; Schalling M; Lander
                    E S; Morgan K; Hudson T J; Richter A
CORPORATE SOURCE:   Montreal Genome Centre, McGill University Health Centre
                    Research Institute, Montreal, Quebec, Canada.
SOURCE:             NATURE GENETICS, (2000 Feb) 24 (2) 120-5.
```

Journal code: 9216904. ISSN: 1061-4036.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
OTHER SOURCE: GENBANK-AB006708; GENBANK-AF193556; GENBANK-AF193557
ENTRY MONTH: 200002
ENTRY DATE: Entered STN: 20000314
Last Updated on STN: 20000314
Entered Medline: 20000228

AB Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS or SACS) is an early onset neurodegenerative disease with high prevalence (carrier frequency 1/22) in the Charlevoix-Saguenay-Lac-Saint-Jean (CSLSJ) region of Quebec. We previously mapped the gene responsible for ARSACS to chromosome 13q11 and identified two ancestral haplotypes. Here we report the cloning of this gene, SACS, which encodes the protein sacsin. The ORF of SACS is 11,487 bp and is encoded by a single gigantic exon spanning 12,794 bp. This exon is the largest to be identified in any vertebrate organism. The ORF is conserved in human and mouse. The putative protein contains three large segments with sequence similarity to each other and to the predicted protein of an Arabidopsis thaliana ORF. The presence of heat-shock domains suggests a function for sacsin in chaperone-mediated protein folding. SACS is expressed in a variety of tissues, including the central nervous system. We identified two SACSmutations in ARSACS families that lead to protein truncation, consistent with haplotype analysis.

L9 ANSWER 3 OF 4 MEDLINE on STN
ACCESSION NUMBER: 1999162199 MEDLINE
DOCUMENT NUMBER: 99162199 PubMed ID: 10053011
TITLE: Location score and haplotype analyses of the locus for autosomal recessive spastic ataxia of Charlevoix-Saguenay, in chromosome region 13q11.
COMMENT: Erratum in: Am J Hum Genet 1999 Apr;64(4):1257
AUTHOR: Richter A; Rioux J D; Bouchard J P; Mercier J; Mathieu J; Ge B; Poirier J; Julien D; Gyapay G; Weissenbach J; Hudson T J; Melancon S B; Morgan K
CORPORATE SOURCE: Service de Genetique Medicale, Hopital Sainte-Justine, 3175 chemin de la Cote Sainte-Catherine, Montreal, Quebec H3T 1C5, Canada.. richtera@ere.umontreal.ca
SOURCE: AMERICAN JOURNAL OF HUMAN GENETICS, (1999 Mar) 64 (3) 768-75.
Journal code: 0370475. ISSN: 0002-9297.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 199904
ENTRY DATE: Entered STN: 19990504
Last Updated on STN: 20000421
Entered Medline: 19990420

AB Autosomal recessive spastic ataxia of Charlevoix-Saguenay (***ARSACS***) is a clinically homogeneous form of early-onset familial spastic ataxia with prominent myelinated retinal nerve fibers. More than 300 patients have been identified, and most of their families originated in the Charlevoix-Saguenay region of northeastern Quebec, where the carrier prevalence has been estimated to be 1/22. Consistent with the hypothesis of a founder effect, we observed excess shared homozygosity at 13q11, among patients in a genomewide scan of 12 families. Analysis of 19 pedigrees demonstrated very tight linkage between the ***ARSACS*** locus and an intragenic polymorphism of the gamma-sarcoglycan (SGCG) gene, but genomic DNA sequence analysis of all eight exons of SGCG revealed no disease-causing ***mutation*** . On the basis of haplotypes composed of seven marker loci that spanned 11.1 cM, the most likely position of the ***ARSACS*** locus was 0.42 cM distal to the SGCG polymorphism. Two groups of ***ARSACS*** -associated haplotypes were identified: a large group that carries a common SGCG allele and a small group that carries a rare SGCG allele. The haplotype groups do not appear to be closely related. Therefore, although chromosomes within each haplotype group may harbor a single ***ARSACS*** ***mutation*** identical by descent, the two ***mutations*** could have independent origins.

L9 ANSWER 4 OF 4 MEDLINE on STN
ACCESSION NUMBER: 93231501 MEDLINE
DOCUMENT NUMBER: 93231501 PubMed ID: 8472930
TITLE: Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Saguenay in northeastern Quebec.

AUTHOR: . De Braekeleer M; Giasson F; Mathieu J; Roy M; Bouchard J P; Morgan K
CORPORATE SOURCE: Departement des Sciences Humaines, Universite du Quebec a Chicoutimi, Canada.
SOURCE: GENETIC EPIDEMIOLOGY, (1993) 10 (1) 17-25.
Journal code: 8411723. ISSN: 0741-0395.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 199305
ENTRY DATE: Entered STN: 19930604
Last Updated on STN: 20000303
Entered Medline: 19930519

AB Autosomal recessive spastic ataxia of Charlevoix-Saguenay (***ARSACS***) is a disorder that has an elevated frequency in Saguenay-Lac-St-Jean (SLSJ) and Charlevoix, two geographically isolated regions in the past of northeastern Quebec. The incidence at birth and the carrier rate in SLSJ were estimated at 1/1,932 liveborn infants and 1/22 inhabitants, respectively, for the period 1941-1985. The mean inbreeding coefficient was twice higher and the mean kinship coefficient 3 times higher among the ***ARSACS*** families than among control families. In the SLSJ region, the birth places of the ***ARSACS*** individuals and their parents did not show a clustered distribution. The genealogical reconstruction suggests that the high incidence of ***ARSACS*** in SLSJ and Charlevoix is likely to be the result of a founder effect. Because the disease is apparently unknown elsewhere in the world and a high proportion of French Canadians presently living in eastern Quebec have ancestors coming from Perche, a small region in France, it also suggests that a unique ***mutation*** accounts for most, if not all, of the ***ARSACS*** cases known in these regions.

=> d his

(FILE 'HOME' ENTERED AT 17:44:28 ON 02 SEP 2003)

FILE 'MEDLINE, CAPLUS, BIOSIS, EMBASE, SCISEARCH, AGRICOLA' ENTERED AT 17:44:48 ON 02 SEP 2003
L1 232 S SPASTIN
L2 30 S SACSIN
L3 257 S L1 OR L2
L4 60 S ARSACS
L5 25 S L4 (P) MUTAT?
L6 12 S L5 (P) L3
L7 5 DUPLICATE REMOVE L6 (7 DUPLICATES REMOVED)
L8 8 DUPLICATE REMOVE L5 (17 DUPLICATES REMOVED)
L9 4 S L8 NOT L7

=> s l8 (p) recombinant
PROXIMITY OPERATOR LEVEL NOT CONSISTENT WITH
FIELD CODE - 'AND' OPERATOR ASSUMED 'L70 (P) RECOMBINA'
PROXIMITY OPERATOR LEVEL NOT CONSISTENT WITH
FIELD CODE - 'AND' OPERATOR ASSUMED 'L72 (P) RECOMBINA'
PROXIMITY OPERATOR LEVEL NOT CONSISTENT WITH
FIELD CODE - 'AND' OPERATOR ASSUMED 'L74 (P) RECOMBINA'
L10 0 L8 (P) RECOMBINANT

=> d his

(FILE 'HOME' ENTERED AT 17:44:28 ON 02 SEP 2003)

FILE 'MEDLINE, CAPLUS, BIOSIS, EMBASE, SCISEARCH, AGRICOLA' ENTERED AT 17:44:48 ON 02 SEP 2003
L1 232 S SPASTIN
L2 30 S SACSIN
L3 257 S L1 OR L2
L4 60 S ARSACS
L5 25 S L4 (P) MUTAT?
L6 12 S L5 (P) L3
L7 5 DUPLICATE REMOVE L6 (7 DUPLICATES REMOVED)
L8 8 DUPLICATE REMOVE L5 (17 DUPLICATES REMOVED)
L9 4 S L8 NOT L7
L10 0 S L8 (P) RECOMBINANT

=> log y
COST IN U.S. DOLLARS SINCE FILE TOTAL

	ENTRY	SESSION
FULL ESTIMATED COST	25.05	25.06

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)	SINCE FILE ENTRY	TOTAL SESSION
CA SUBSCRIBER PRICE	-1.95	-1.95

Connection closed by remote host